Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Spinocerebellar Ataxia Autosomal Recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354.

Thursday, 25 May 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Anthony Rutherford Bobbie Farsides	
Members of the Executive	Bernice Ash Paula Robinson	Secretary Head of Planning & Governance
External adviser	Dr Jenny Carmichael	
Legal Adviser	Sarah Ellson	Fieldfisher
Observers		

Declarations of interest

- Members of the panel declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 8th edition of the HFEA Code of Practice
- Standard licensing and approvals pack for committee members.

The following papers were considered by the committee:

- Executive Summary
- PGD Application Form
- Redacted peer review
- Genetic Alliance opinion
- Minutes of three previous Statutory Approval Committee decisions

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Jenny Carmichael, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Spinocerebellar Ataxia 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient perspective and supported the application.

1.4. The committee had regard to its decision tree. The committee noted that the centre is licensed to carry out PGD. The committee was also satisfied that the centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.

1.5. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.6. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e. 'where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality'.

1.7. The committee noted that the condition is inherited in an autosomal recessive manner which means there is 25% chance of having an affected child in each pregnancy, if each parent has a relevant mutation.

1.8. This recessive condition causes moderate to severe (usually severe) intellectual disability and speech is absent or in some cases impaired. Tone is reduced. Walking is severely delayed. Some teenagers/adults have been reported who can only crawl, therefore requiring a wheelchair. Affected people are ataxic, therefore have a difficulty with voluntary coordination of muscle movements and may have scolioisis (curvature of the back). The majority have sensory neural hearing loss. Penetrance is thought to be 100%.

1.9. The committee noted no treatment is available, although supportive strategies may be applicable to manage individual symptoms of the condition, for example, wheelchair use. Seizures may be manageable with anti-convulsant medications. Quality of life is reduced by both physical and intellectual disability. Many affected individuals will be fully dependant throughout their lives for all activities of daily living.

1.10. The committee noted that the condition applied for is Spinocerebellar Ataxia 20, SCA20 (SNX14-related cerebellar hypoplasia), OMIM #616354. On the OMIM website, OMIM #616354 already corresponds to spinocerebellar ataxia, autosomal recessive 20; SCAR20. The inspectorate notes the presence on the OMIM website of another condition: Spinocerebellar Ataxia 20, SCA20, OMIM # 608687, which is a gene duplication disorder. To prevent confusion, it is suggested that the condition be added to the HFEA website as Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354.

1.11. The committee noted the inspectorate's recommendation to consider the approval Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354 to be included on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354 is serious given the early onset and life-changing effects of this condition, including severe intellectual impairment, inability to communicate, seizures and the lack of a cure. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

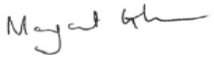
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing of embryos for Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354 and agreed that the condition should be included on the PGD List on the HFEA website as Spinocerebellar Ataxia autosomal recessive 20, SCAR20 (SNX14-related cerebellar hypoplasia), OMIM #616354.

3. Chair's Signature

3.1. I confirm this is a true and accurate record of the meeting.

Signature



Name

Margaret Gilmore

Date

8 June 2017